The fetal abdominal wall defects using 2D and 3D ultrasound. Pictorial essay

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Abstract

The use of fetal ultrasound in prenatal care allows the diagnosis of the majority of abdominal wall defects with subsequent opportunities for parental counseling and optimal perinatal management. Outcome of a fetus with an abdominal wall defect is significantly affected by the presence of additional malformations or chromosomal defects. Appropriate management is dependent on the early identification of such anomalies in addition to accurate delineation of the abdominal wall defect itself. Cases with anterior wall defects are presented to illustrate the spectrum of appearances with 2D and 3D ultrasound. 2D combined with 3D ultrasound are very effective methods in the diagnosis of these anomalies and MRI should be reserved only for difficult and intricate cases.

Keywords: ultrasonography, omphalocele, gastroschisis, body stalk anomaly, Cantrell pentalogy, bladder extrophy

Abdominal wall defects comprising gastroschisis, omphalocele, Cantrell pentalogy, bladder extrophy, and limb body wall complex, remain a source of significant morbidity and mortality, despite the advances in neonatal and pediatric surgical care. The correct prenatal diagnosis is extremely important for patient management. The key feature for ultrasound when distinguishing these conditions is the position of the defect in relation to the umbilical cord insertion. We performed a systematic review of the most important features in both 2D and 3D ultrasound of the abdominal wall defects. The paper, along with its graphics, is meant as a visual testimony of the fact that both 2D and the 3D ultrasound represents very useful tools for the diagnosis of these pathology.

Omphalocele is an anterior abdominal defect at the base of the umbilical cord, with herniation of the abdom-

inal contents. The incidence of the condition is one in 4000-7000 live births [1]. Omphaloceles are associated with other anomalies in more than 70% of the cases, most of which are chromosomal [2].

The etiology of omphalocele is not known. The various theories that have been postulated include failure of the bowel to return into the abdomen by 10-12 weeks, failure of lateral mesodermal body folds to migrate centrally and persistence of the body stalk beyond 12 weeks of gestation [1]. Physiological umbilical herniation occurs during the eighth week of development when the fetal midgut extends into the extraembryonic celom, occupying the proximal segment of the umbilical cord. At 8-10 weeks of gestation, all fetuses express physiological umbilical herniation of the midgut (fig 1).The midgut returns to the abdominal cavity at 12 weeks of menstrual age. A physiological hernia seldom exceeds 7 mm in diameter or rarely persists after 12 weeks of gestation (fig 2). Due to this physiological herniation the diagnosis of omphalocele should be established after 12 weeks of amenorhea. Omphalocele is more likely to be associated with multiple anomalies and have a chromosomal or genetic syndrome etiology when compared to other abdominal wall defects such as gastroschisis. In many cases (45-88%) ompha-

Fig 1. Longitudinal section of an embryo at 9 weeks demonstrating the physiological umbilical hernia

Fig 2. 3D image of an embryo at 12 weeks – physiological hernia is not visible anymore

Fig 3. 3D image an of embryo at 12 weeks and 5 days with omphalocele

Fig 4. Longitudinal section of an embryo at 14 weeks and 2 days with omphalocele

Prenatal testing is available by ultrasound and maternal serum α-fetoprotein. The sensitivity of ultrasound screening is estimated at 80% [1]. Ultrasonography (USG) is the imaging modality of choice for the prenatal assessment of the foetus. The earliest moment an omphalocele can be detected is at 12 weeks of menstrual age (fig 3, fig 4).

An omphalocele is diagnosed on USG when a fetal anterior midline mass consisting of abdominal contents that have herniated through a midline central defect at the base of umbilical insertion is demonstrated (fig 5, fig 6). This mass has a smooth surface represented by peritoneal-amniotic membrane. The umbilical cord enters this mass and the umbilical vein is present within the omphalocel (fig 7).

The size of the exomphalos can vary, from a small umbilical hernia (hernia into the cord) to a large abdominal wall defect with extrusion of all abdominal viscera (giant omphalocele).The mean size of the defect is 2.5-5 cm, with fetal ascites [4]. In cases with difficulty to identify the exact pathology and to demonstrate the co-existing pathologies, 3D ultrasound may be helpful. Besides its help as a diagnostic tool, 3D ultrasound may help the family to better understand the condition (fig 8, fig 9). The size is appreciated sonographically by measuring the ratio of the transverse diameter of the omphalocele over to the transverse diameter of the abdomen. It is assumed that if this ratio is more than 60% it can contain the liver, and if is less it usually contains the bowels. The identification of hepatic vessels inside the herniation sac could be done with Doppler examination. Sometimes the whole liver is seen outside the abdomen and in this case the thoracic and abdominal cavity could be small and underdeveloped. Polyhydramnios, and occasional oligohydramnios with associated anomalies are common [5]. There are some phonographic hints regarding the possibility of a chromosomal anomaly. It

locele is associated with different anomalies and its severity determines the prognosis [1]. The most common anomalies are cardiac, genitourinary, gastrointestinal, musculoskeletal, neural tube and head neck defects. The chromosomal anomalies are present in 40-60% of fetuses with exomphalos and include trisomies 18,13 and 21, Turner, Klinefelter and triploidy syndromes [3].

Fig 5. Longitudinal of the abdomen with ompha-locele. The defect is in the midline with umbilical cord inserted to the top of the protruding mass.
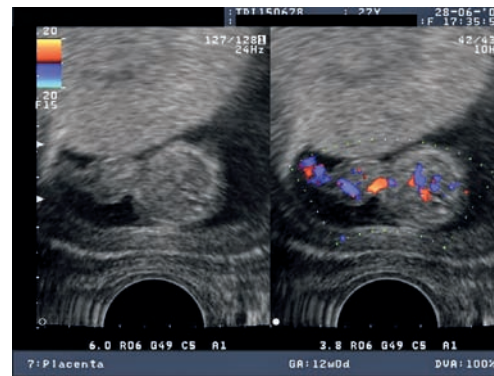

Fig 7. A transverse view of the abdomen with om-phalocele. Umbilical vein is present within the om-phalocel.

Fig 6. Another case with omphalocele – longitudi-nal view of the body.
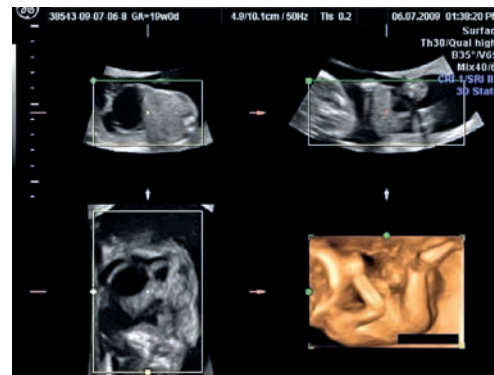

Fig 8. 3D ultrasound of a large omphalocele.

is thought that a chromosomal anomaly is more likely when oligohydramnios or polyhydramnios complicates the pregnancy. Chromosomal anomalies are more com-monly associated with omphalocele that contains only the bowel compared to those that contains only the liver or bowel and liver [2]. Whether these signs are present or not, the detection of an omphalocele obliges a chro-mosomal detection.

When the diagnosis of omphalocele is established a careful examination of the fetus should be done in order to detect if there are associated anomalies [6]. The most frequent associated anomalies are cardiac, gastrointesti-nal, genitourinary, and neural tube defects.

Differential diagnosis of an omphalocele include gas-troschisis, amniotic band syndrome, bladder exstrophy, pentalogy of Cantrell, body stalk anomaly, pseudoom-phalocel, acardiac monster. The mortality rate is 80% when associated anomalies are present and it increases to 100% when chromosomal and cardiovascular anomalies are present [7]. One recent study has shown that peri-

Fig 9. 4D rendering of the omphalocele – Sagittal 3D view showing the omphalocele at 16 weeks

conceptional multivitamin use is associated with a 60% reduction in the risk of nonsyndromic omphalocele [8].

Gastroschisis, is the herniation into the amniotic cavity of abdominal contents through the abdominal wall without involving the umbilical cord. The prevalence of

gastroschisis is 1 in 10.000 [1]. The term gastroschisis is derived from Greek roots meaning "stomach cleft", but the correct term may be laporoschisis (belly cleft) [9]. Chromosomal anomalies are very rare.

The abdominal wall defect in the fetus permits the small and large bowel to herniate through the defect and be exposed to the amniotic fluid environment. This herniation protrudes into the amniotic cavity usually in the right paraumbilical region. No covering membrane is present. The defect is usually small, less than 4 cm in diameter through which various segments of bowel and other abdominal organs may be herniated. Gastroschisis can produce intrauterine and neonatal complications, which may include postnatal bowel dysfunction, bowel atresia, bowel necrosis and subsequent short bowel syndrome [2]. Gastroschisis is thought to result from a vascular event of the omphalomesentric artery. Affected patients have a malrotated bowel. Vascular compromise may occur from a volvulus, and it may result in bowel obstruction, ischemia or atresia. Besides malrotation, gastroschisis is very rarely associated with other anomalies- congenital heart disease, ectopia cordis, neural tube and diaphragmatic defects. When it is associated with other anomalies is it possible to be a ruptured omphalocele [1].

Routine ultrasound has allowed this birth defect to be identified in utero with high specificity and sensitivity. Prenatal diagnosis by ultrasound is based on the visualization of a herniated free-floating bowels without any membranous and a normal umbilical cord (fig 10, fig 11). Serial ultrasound is mandatory because of the risks which could occur: bowel obstruction, peritonitis, bowel perforation and IUGR. Seventy percent of infants with gastroschisis are below the 50th percentile in weight [1]. In order to estimate the risk of postnatal bowel complication the bowel diameter should be measured. A diameter greater than 11 mm is a sign for postnatal complications [4]. The anomaly can be diagnosed as early as 12 weeks of gestation. There are some reports in the literature of an earlier diagnosis [10,11].

Intra-abdominal bowel dilatation is found more often on prenatal ultrasound in fetuses with gastroschisis that develop bowel obstruction requiring surgery after birth. This information may be useful for identifying those fetuses with gastroschisis that are more likely to have a significant bowel obstruction after birth [12]. The prevalence of intrauterine growth restriction is increased in infants with gastroschisis but is overestimated with prenatal ultrasound due to smaller abdominal circumference measurements [13]. Fries et al reported no difference in the postnatal outcome for those growth-retarded infants compared to non-growth-retarded infants with gastro-

schisis, but the lack of a statistically significant difference is probably a reflection of the small population size with 10 growth-restricted and 11 non-growth-restricted infants [14].

Differential diagnoses include physiologic bowel herniation, omphalocele, amniotic band syndrome, bladder and cloacal extrophy, body stalk anomaly, cavernous hemangioma, pentalogy of Cantrell [1,4]. The prognosis is favorable. Postoperative survival is about 92% if the gastroschisis has no other associated anomalies [15].

The Limb Body Wall Complex (LBWC) is a rare, polymalformative fetal syndrome with the three essential features: exencephaly/encephalocele and facial clefts, thoraco- and/or abdominoschisis and limb defects [16]. Generally, the diagnosis is based on two or three of the above features. A constant finding is celosomia which can be variably associated with encephalic, vertebral, visceral or limb anomalies. The incidence of this uncommon disorder is 0.21-0.31/10.000 live births [17]. Some authors consider that LBWC simply represents a severe
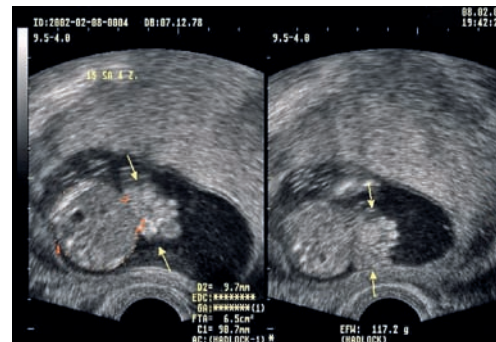
Fig 10. Pregnancy of 23 weeks with gastroschizis - bowel loops are seen to exit the abdomen through a defect above and to the right of a normal cord insertion. No membrane covers these bowel loops.

Fig 11. Gastroschizis – free-floating loops of small bowel at 23 weeks of gestation.

form of amniotic band syndrome. This is sustained by the presence of amniotic bands in almost 40% of cases. The pathogenesis of LBWC is unclear and uncertain. Recently, Hunter et al. launched a new theory which sustains that the association of malformations originates as early as the embryonic disc stage and that some of the associated anomalies are secondary complications of the primary disturbance in embryo genesis [18].

The diagnosis of LBWC is made by the combination of a large abdominal wall defect with protrusion of the viscera, a severe spinal scoliosis and a continuous juxtaposition of the fetus to the placenta. The eviscerated organs form a complex mass entangled with membranes. The defect comprises most often both the abdomen and the thorax or rarely can be limited only to one of them (fig 12). Associated malformations included central nervous system lesions, facial abnormalities, cardiac malformations, urogenital anomalies, limb defects, amniotic bands and placental abnormalities. The limb defect has different forms: clubfoot, oligodactyly, arthrogryposis,

Fig 12. The Limb Body Wall Complex – longitudinal view of dysmorphic abdominal contents with no detectable abdominal wall.

Fig 13. The Limb Body Wall Complex – scoliosis of the thoracic spine.

absent limbs or digits. Severe scoliosis develops as a consequence of the irregular attachment of the fetus to the placenta (fig 13) [19].

Russo et al. identified two distinct phenotypes of LBWC: placento-cranial and placento-abdominal types [20]. The placento-cranial type is characterized by craniofacial defects, facial clefts, and amniotic adhesion. The placento-abdominal type has no cranial defects, but is more frequently associated to celosomia, lumbosacral meningomyelocele and kyphoscoliosis and urogenital anomalies.

LBWC is usually diagnosed during the second trimester of pregnancy by ultrasound. Although difficult because of the physiological herniation of the bowel, the syndrome can also be diagnosed in the first trimester of pregnancy [21,22]. An ultrasound examination correctly performed can diagnose early this condition earlyand enable the possibility of performing a therapeutic abortion. It's important to distinguish body stalk anomaly from other anterior wall defects to determine the management options. The prognosis of LBWS is very poor and because it is considered that LBWC is incompatible with life, the pregnancy should be terminated after a correct sonographic diagnosis. Although the great majority of cases described in the literature are in the late second or third trimester of pregnancy, an early diagnosis is possible. It is also important to explain to the families affected that there is no recurrence risk in this anomaly.

The **pentalogy of Cantrell** is a rare congenital defect involving the abdominal wall, sternum, diaphragmatic pericardium and the heart. The first description of the syndrome was made by Cantrell and Haller in 1958 [23]. The syndrome occurs with various degrees of severity from incomplete to severe expression with or without involvement of other organ systems. The estimated incidence of Cantrell pentalogy is 5.5 per 1 million live births [24].

In the Cantrell pentalogy the sternum defects may vary from a simple notch of the manubrium to absence of the entire sternum. Abdominal wall defect is represented by omphalocele, diastasis recti, epigastric hernia, umbilical hernia, and combined defects. The most common of these are omphalocele. Ventral defects of the diaphragm and absent pericardium are the most common diaphragmatic and pericardial defects, respectively (fig.14). Ectopia cordis is characterized by complete or partial displacement of the heart outside the body. The most common intracardiac defects as described by Cantrellll are ventricular defects and atrial defects followed by tetralogy of Fallot and left ventricle diverticulum. Together with the congenital defects described by Cantrell in 1958 many cases have associated different anomalies, many of them worsen the prognosis**.** Additional anomalies include: craniofacial defects (cleft lip and/or palate),

Fig 14. Pentalogy of Cantrell with eventrated abdominal content of the fetus at 14 weeks of amenorheea.
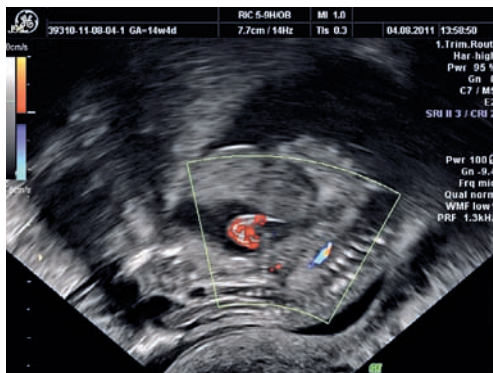

Fig 15. Color Doppler sonogram in a patient with pentalogy of Cantrell Power Doppler image showing ectopic heart of the fetus.
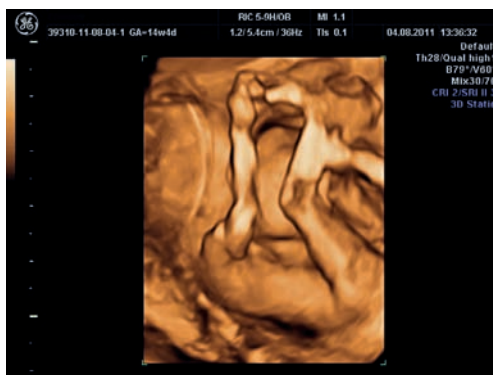

Fig 16. 3D ultrasound of the Cantrell pentalogy.

central nervous system (encephalocele, hydrocephalus and craniorachischisis), and limb defects such as club-foot, absence of tibia or radius and hypodactily.

The pentalogy of Cantrell can be diagnosed with ultrasound beginning the first trimester of pregnancy

[25]. The ultrasound reveals the association between an omphalocele and an ectopic heart. A very useful exam is Power Doppler which visualizes the heart outside the thorax (fig 15). Pleural and pericardial effusions are common and some authors consider them as indirect markers for the pentalogy [26]. It is important to notify that there is the possibility that some sonographic aspects could improve during the course of pregnancy [27]. The diagnosis in the first trimester of pregnancy should be made after 12 weeks of gestation because of herniation of bowel out of the abdomen is a normal event in fetal development until this time. Although 3D is useful in confirming the diagnosis it is not mandatory but it provides a complete view of the anomaly prenatally (fig 16). The early diagnosis in the first trimester is possible using ultrasound and is desirable because it offers the option of arresting the pregnancy progress.

The prognosis will vary according to the severity of the anomalies. Few patients survive, and even fewer survive with good outcomes of quality of life. The prognosis is poorer in the complete form of pentalogy and in cases with other anomalies associated [28].

Bladder extrrophy is rare, occurring with an incidence of 1 per 30,000–50,000 live births. [29]. It is characterized by incomplete closure of the lower anterior abdominal wall and exposure of the urinary bladder into the amniotic cavity. Associated genitourinary findings include extension of the bladder defect into the urethra, incomplete testicular descent, and bilateral inguinal hernias. Bladder extrophy is also associated with OEIS complex (omphalocele, bladder extrophy, imperforate anus, spinal defects).

The prenatal ultrasound findings of bladder exstrophy are: the presence of a solid bulging mass in the lower abdominal wall, non-visualization of the urinary bladder and normal amniotic fluid volume [30,31]. Minor findings are low insertion of the umbilical cord, widening of the iliac crests, normal kidneys, small penis and epispadia [32].

The differential diagnosis includes all other anterior abdominal wall defects such as omphalocele, gastroschisis and cloacal exstrophy. In the case of the first two defects, there is an extruded sac or herniated bowel, but a normally filled bladder should be seen in the pelvic cavity. In the case of omphalocele, the umbilical arteries may run inferior to the mass and the umbilical vein runs through the herniated liver. Depiction of the umbilical vein within the bulging mass prompts the diagnosis of omphalocele. The diagnosis of cloacal exstrophy is often difficult but can be made when more complex anomalies involving the gastrointestinal tract and spine are involved. Prognosis is good for bladder exstrophy, with surgical intervention required for primary closure or excision with urinary diversion.

Conclusion

The use of fetal ultrasound in prenatal care allows the diagnosis of the majority of abdominal wall defects with subsequent opportunities for parental counseling, fetal intervention, and optimal perinatal management. 2D and 3D ultrasound allow an early and correct diagnosis of these types of anomalies. Cases diagnosed with abdominal wall defects should be considered for in utero transport to perinatal center in order to benefit from an optimal management with a multidisciplinary team. In cases with associated lethal or multiple severe abnormalities, parents may opt for elective termination of the pregnancy.

Conflict of interest: None

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